GTR: Genetic Testing Registry

Central repository of genetic tests voluntarily supplied by test providers

https://www.ncbi.nlm.nih.gov/gtr

National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

Scope

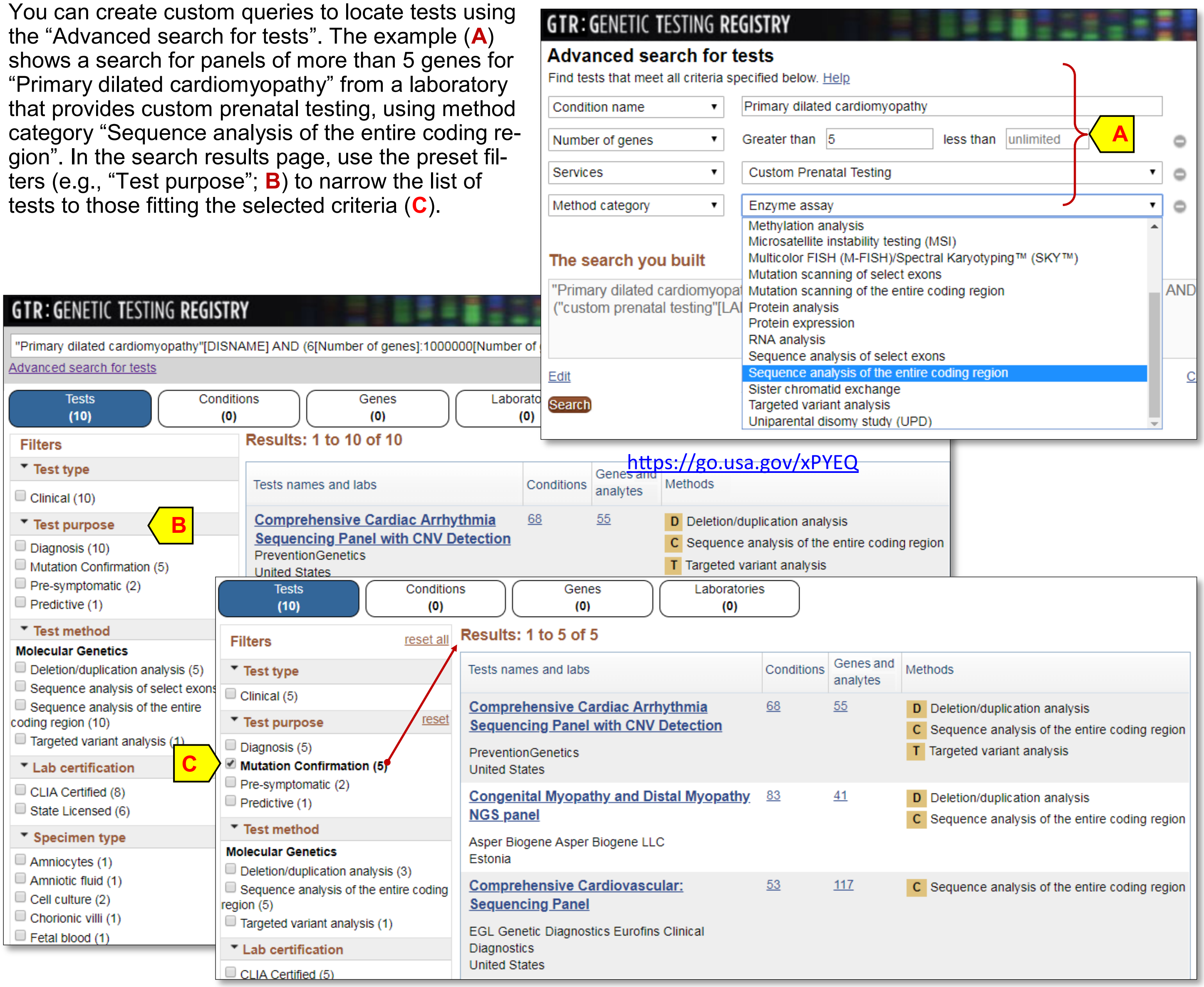
The Genetic Testing Registry (GTR) is a free online resource that provides centralized access to comprehensive genetic test information voluntarily submitted by test providers. GTR covers clinical and research tests for heritable and somatic mutations, including pharmacogenetic tests as well as tests using complex arrays and multiplex panels. GTR provides a wide range of information, such as, ordering information, purpose of the test and its limitations, testing method(s), and what the test measures. Clinical tests have information on analytical validity, as well as evidence of clinical validity and clinical utility. Research tests have information on the study and participation requirements. The name, location, contact information, and credentials of laboratories are displayed. GTR records provide links to context-specific information about conditions, genes, test standards, practice guidelines, and consumer support sites. The primary audience of GTR is the health care community.

Data Access

The GTR homepage (right) allows the retrieval of information through several different search strategies using tabs above the search box (**A**). You can search by the test name, disease, drug response, gene symbol, laboratory name, director and staff names, and laboratory location. A tab for *GeneReviews*, containing reviews on over 700 conditions, enables searching of this key resource directly from the GTR homepage. A set of YouTube video tutorials (**B**) provides a quick guide on how to search for and submit information to GTR. Links to GTR documents are in the "About GTR" panel (**C**). The homepage also provides a summary of participation by laboratories and data growth trends (**D**). Links to locate a genetics professional, as well as genetics resources incorporated in GTR (**E**) are provided. The submission section (**F**) provides links to the submission interface along with help documentation.

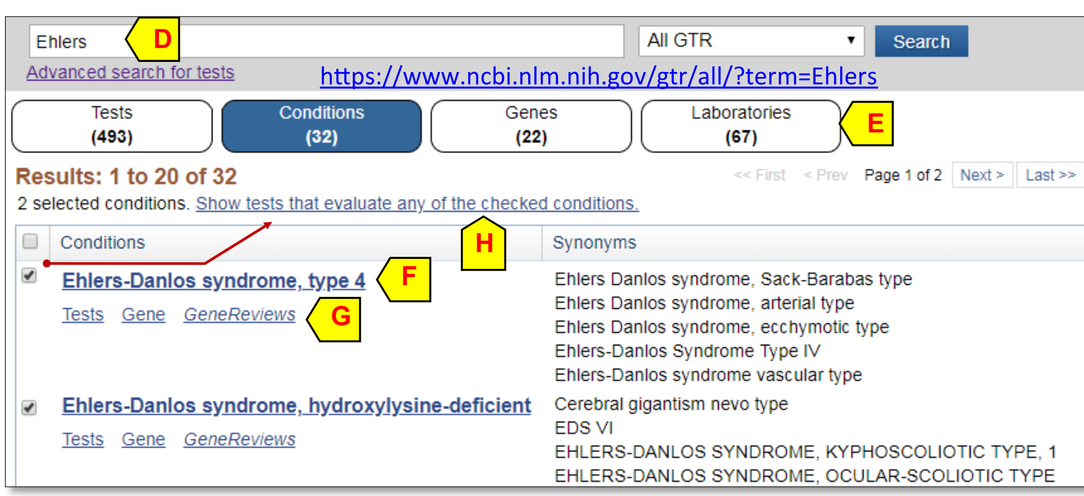
Using Advanced Search to Find Specific Tests

You can create custom queries to locate tests using the “Advanced search for tests”. The example (**A**) shows a search for panels of more than 5 genes for “Primary dilated cardiomyopathy” from a laboratory that provides custom prenatal testing, using method category “Sequence analysis of the entire coding region”. In the search results page, use the preset filters (e.g., “Test purpose”; **B**) to narrow the list of tests to those fitting the selected criteria (**C**).



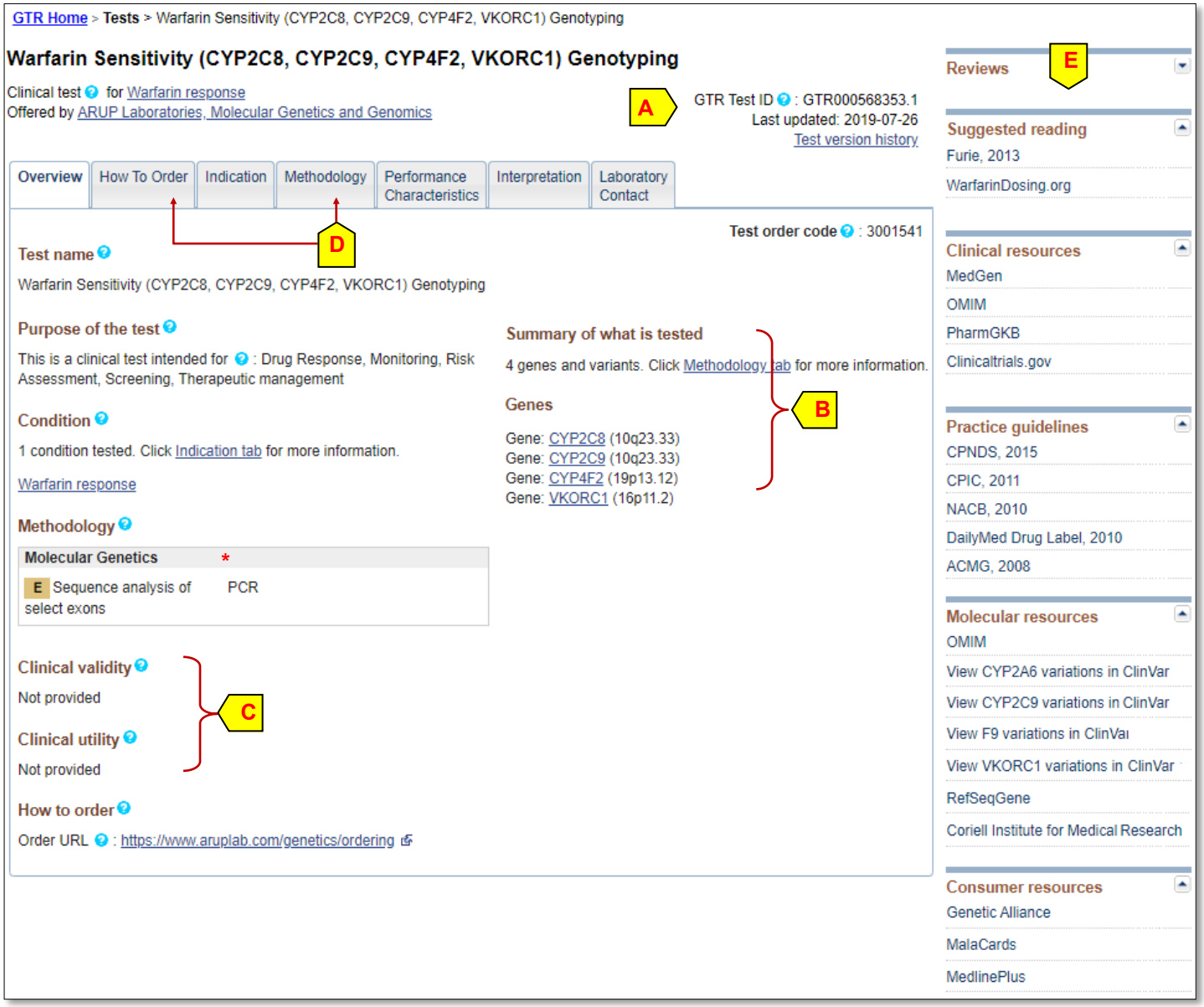
Using the ‘All GTR’ Tab

On the GTR home page, the main search box is preset to the “All GTR” tab. Enter a term (e.g., “Ehlers”) and select from the suggested dropdown list to go to the specific page or click the “Search All GTR” button (**D**) to get a list of results where you can click the corresponding buttons to access Tests, Conditions, Genes, and Laboratories (E**B**). On the Conditions results page, the name of each condition (**F**) links to the page with condition-specific information (e.g., clinical features, practice guidelines) with links below to available tests, associated genes, and article(s) from *GeneReviews* (G**D**). Checking the boxes for any conditions activates a link (**H**) at the top, which can be used to retrieve a subset of tests for any of the selected conditions.



Information in a Genetic Test

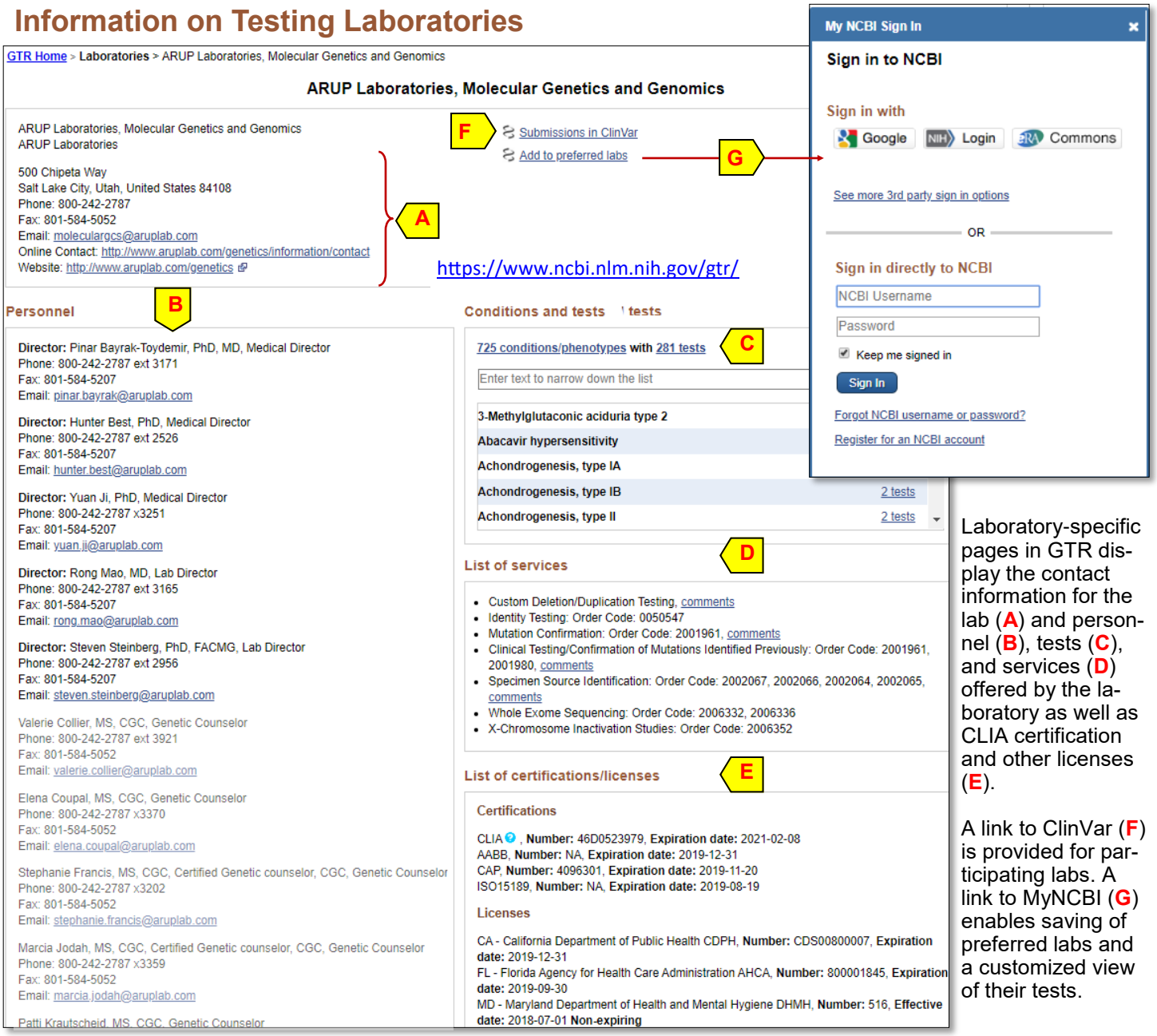
The example shows a GTR-registered test for warfarin response (https://www.ncbi.nlm.nih.gov/gtr/tests/568353) identified by a stable accession and version (**A**). The test information page opens to show an overview of the test, including the **Purpose of the test**, a **Summary of what is tested**, the **Condition**, and **Genes** involved (**B**), as well as reported **Clinical Validity** and **Clinical Utility** if relevant and provided (**C**).



Other information is provided in tabs (**D**) such as **How to Order** and **Methodology**, which includes the methods and targets that the test interrogates.

Links to important resources (**E**) are shown on the right. They include **Clinical resources** from NCBI (MedGen and Clinicaltrials.gov) and elsewhere, **Practice guidelines** from professional organizations, as well as **Molecular resources** from NCBI and some outside organizations.

Information on Testing Laboratories



Laboratory-specific pages in GTR display the contact information for the lab (**A**) and personnel (**B**), tests (**C**), and services (**D**) offered by the laboratory as well as CLIA certification and other licenses (**E**).

A link to ClinVar (**F**) is provided for participating labs. A link to MyNCBI (**G**) enables saving of preferred labs and a customized view of their tests.

Submit Test information to GTR

GTR accepts submissions for laboratory data and clinical and research tests. You will need a MyNCBI account. Detailed instructions are available at www.ncbi.nlm.nih.gov/gtr/docs/submit/. Submitters may choose to provide their data via:

- Online forms
- Spreadsheets for bulk submissions of clinical tests, either fully automated or semiautomated
- Fully automatic XML submission

GTR is designed to minimize burden on submitters, with features such as menus, “type ahead” functionality, and text fields to allow cut-and-paste of information. Where possible, fields are automatically populated for the submitter. In addition, test data identical for all tests in the lab’s menu can be entered in the “Default parameters” section of the lab record to pre-populate fields for new tests being registered.

GTR provides information about the test provider as well as the availability, accuracy, validity and usefulness of each test; therefore, a minimal set of fields are required for all submissions. For the complete list of required and optional fields, see www.ncbi.nlm.nih.gov/gtr/docs/fieldrequirements/. Research tests additionally require information about the study and researchers.

Contact

GTR welcomes your feedback. Please sendquestions or comments to: gtr@ncbi.nlm.nih.gov